

SHEET 1 OF 12

Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS

Inventors: Stone *et al.* Ref. No. 21087.0019U3

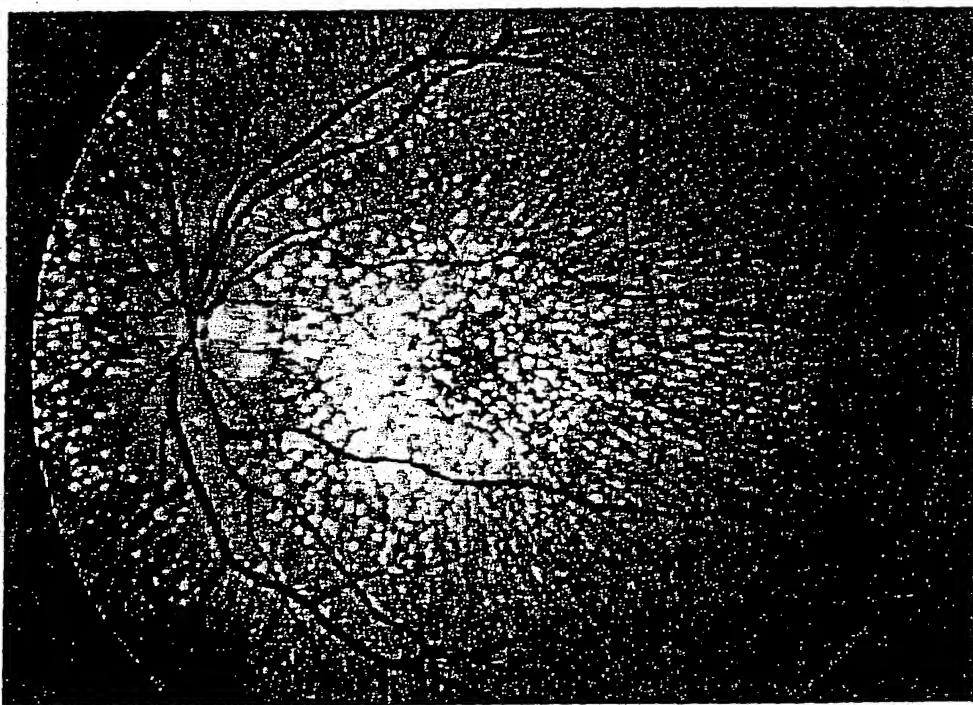


FIG. 1

SHEET 2 OF 12

Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS  
Inventors: Stone *et al.* Ref. No. 21087.0019U3

p-tel  
D2S119  
D2S2227  
D2S2316  
SGC-31133  
WI-11560  
D38521  
stSG10198  
D2S2251  
SGC-35022  
D2S2352  
BCD1993  
SGC-34889  
WI-22280  
RP-S27A1  
WI-1332  
A009M26  
SGC-32447  
D2S2153  
WI-15186  
WI-16857  
133O18AAAT  
B-FODRIN 5'  
B-FODRIN 3'  
293J12CA  
133O18CA  
FBNL INTRO 2  
FBNL EXON 7  
FBNL EXON 9  
FBNL EXON 10  
FBNL INTRON 11  
322A4-33  
322A4 AAAT  
WI-6613  
322A4-29  
WI-6704  
SGC-34452  
WI-6241  
WI-18292  
stSG-42480  
D2S378  
SGC-31346  
WI-11399  
D2S1364  
SGC-34988  
D2S357  
D2S147  
cen

14cM ML RECOMBINATION INTERVAL, HEON, *et al.*, 1996

5 cM DHHD RECOMBINATION INTERVAL, GREGORY *et al.*, 1996

6.2 cM ML RECOMBINATION INTERVAL, EDWARDS *et al.*, 1998

INITIAL SWISS HAPLOTYPIC INTERVAL

AUSTRALIAN RECOMBINATION INTERVAL

FINAL SWISS HAPLOTYPIC INTERVAL

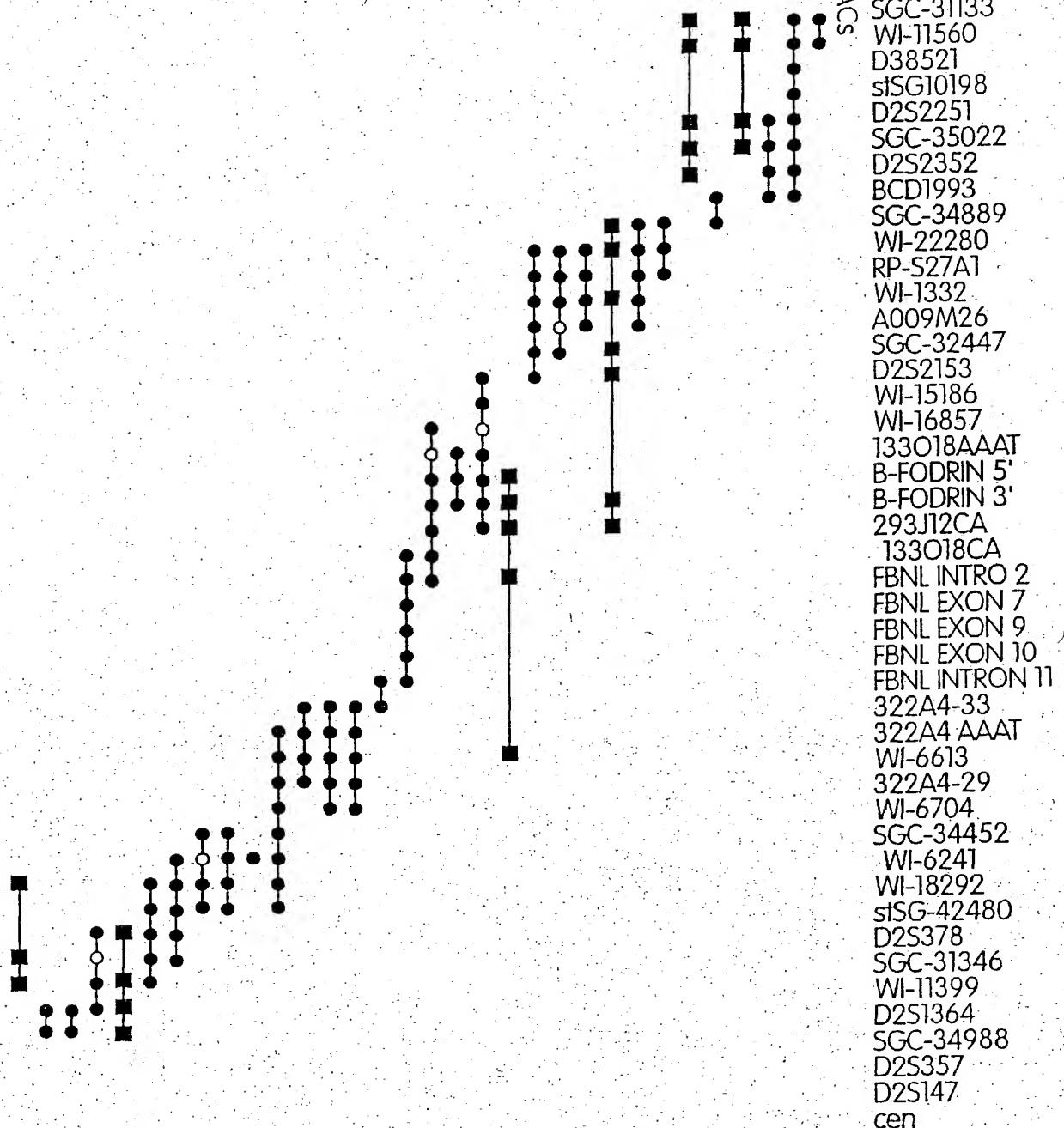
FIG. 2A

## SHEET 3 OF 12

Title: MACULAR DEGENERATION  
 DIAGNOSTICS AND THERAPEUTICS  
 Ref. No. 21087.0019U3

Inventors: Stone *et al.*

BACs and YACs  
 113D22  
 222A14  
 347N1  
 885D2  
 223J15  
 769G5  
 154E5  
 543A16  
 919A6  
 33E15  
 483L9  
 560B19  
 909E11  
 270H2  
 133018  
 293J12  
 223J15  
 582N15  
 670023  
 319E10  
 322A4  
 377013  
 188B24  
 566D20  
 496F2  
 343B24  
 320E2  
 834C1  
 202J21  
 432P15  
 915H2



p-tel  
 D2S119  
 D2S2227  
 D2S21316  
 SGC-31133  
 WI-11560  
 D38521  
 stSG10198  
 D2S2251  
 SGC-35022  
 D2S2352  
 BCD1993  
 SGC-34889  
 WI-22280  
 RP-S27A1  
 WI-1332  
 A009M26  
 SGC-32447  
 D2S2153  
 WI-15186  
 WI-16857  
 133018AAAT  
 B-FODRIN 5'  
 B-FODRIN 3'  
 293J12CA  
 133018CA  
 FBNL INTRO 2  
 FBNL EXON 7  
 FBNL EXON 9  
 FBNL EXON 10  
 FBNL INTRON 11  
 322A4-33  
 322A4 AAAT  
 WI-6613  
 322A4-29  
 WI-6704  
 SGC-34452  
 WI-6241  
 WI-18292  
 stSG-42480  
 D2S378  
 SGC-31346  
 WI-11399  
 D2S1364  
 SGC-34988  
 D2S357  
 D2S147  
 cen

FIG. 2B

SHEET 4 OF 12

Title: MACULAR DEGENERATION  
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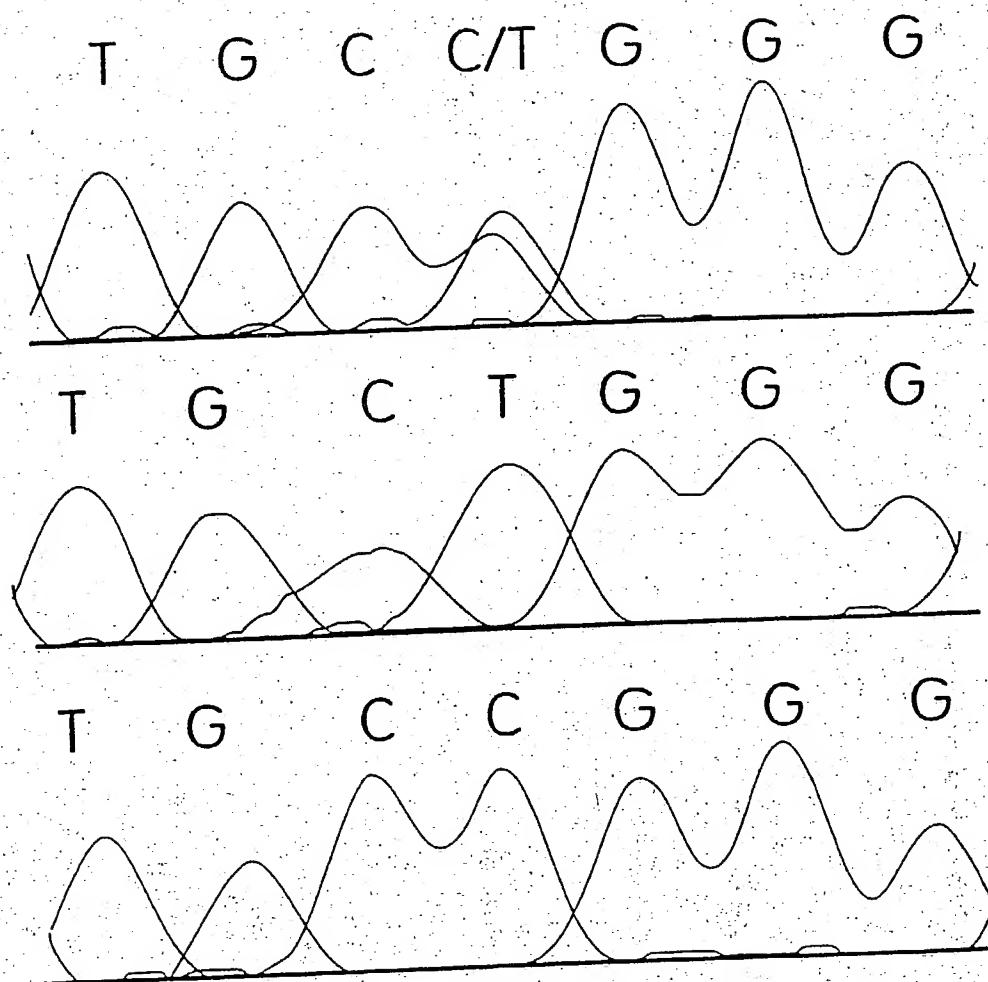


FIG. 3

SHEET 5 OF 12

Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS  
Inventors: Stone *et al.* Ref. No. 21087.0019U3

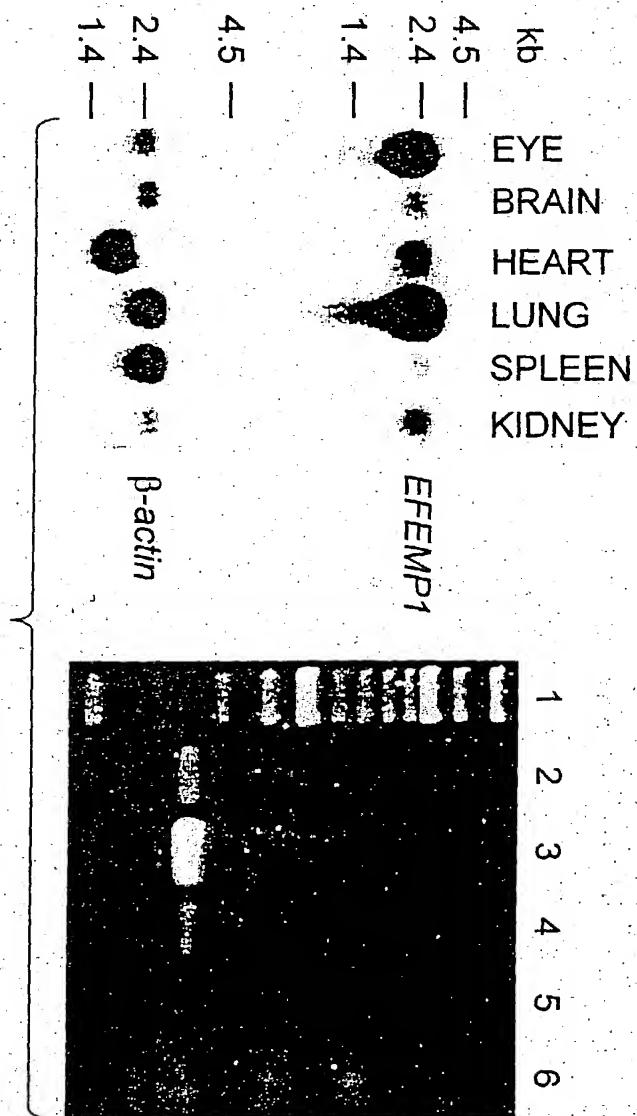


FIG. 4

SHEET 6 OF 12

**Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS**

### Exon 1

Inventors: Stone *et al.* Ref. No. 21087.0019U3

Currently undetermined

exon 2 partial transcript

AGTGTGCGAGATTAAGCCGACCTGGATTCCATAGGAGCTGGTTAGAAGCTGGGACGCTGAGCAGCT  
CCAGGGGACCGCCGCGTTAGCTTGTGTTAAGAAAGGGGACCTCATCTCCCTGCCGGGCCAGGCCGCC  
CGCCCCGAAACTGGTACCTTGGGCTCGCGGTGCGATCCCTGGTTCGGTCCTAGGCAGCCTGAAACGAA  
GGTAGCGTGTGGGGACCCAGACTGATAAGACAAAAGAGAATCAGTCGCTTGGGCTGCCCTCCACA  
CAACCTGGGACTTTAAACAAAGCTGTGCGAGAGAAAGGCGTGGAAATGCCACTTTGAGAGTTGTG  
CTGGGGGATGTGAGAAGCTGAGACATGTGAGAAGGTCTAGTATTCTACTAGAACTTGGAGAGATTGCT  
CTCCGAGTTTGTATTGTAAAGGCTTGAGGCAAGGCAATTATTCATATTGGCT  
CACAGGTATTTGTGTGCTGCAAGGAACCTGCTAGCTCAAG (SEQ ID No. 46)

### Complete intron 2

### Exon 3

ATTACACAATG TTGAAAGCCC TTTTCCTAAC TATGCTGACT CTGGCGCTGG TCAAGTCACA  
GGACACCGAA GAAACCATCA CGTACACG (SEQ ID No. 48)

Start intron 3

gttaagggtt atggaatttg aaataagtac ggttcagcgg gattctgtgacaaaaaaaaaaga ctttacacac  
ccctgcagt gtatgggc tatactctgtgagggtgat aaattaaaca acacttcatt catgcttcat  
atctaagattcggtttaat tgcccccttg atccttcaa aagttcattt ggctcaccacctaagatagg  
aaccacatg taatcatttgcaggctaaatggatccgttcaaaa actaaaacca aagaaaggta  
catgtttcca aaacattcaacaaattaatg ggtgtaaaggactggaaaac ctggactct  
accacatgcagataaaacca atacgtgcag aataagactc aagtcaagta agaacgttacaccataaa  
gacacatggccttcttgcgtacatgacatgcatttcaagtaagtggccttattgaatttataaaggctata  
tattcattttttgtataactt gataattctaataaataaaggcagacaacagttatgtgttaccaggatg  
catattggctaaagtggttttaaaacgtaatgtgtgcaactccgtttgcatttctaattgcgtct  
ctgatatttccaagtaatattgatttagttactgtgtcataggtgtaccatgtttaataaaatattaaaaagat  
cacctgaccctccactgtcataaaatagttgtggtagaaac agagaaggacagactgacttcacttctgt  
gagttgttgcacctctgtttctgttttcaatcagtgttaggcaatgacattgtcctggatt  
ggaatatgaaaa gcacattttctactgtcccgatttaaaattaatgacttactcactcgaaagaatgtgaaa  
aattttgaaaagaaaactctaaaaatgacttactgtcaattactgataataaac attatctcaacttttgg  
taccacattatccctagaatgttagtattcatctggcaatgttccttctgttgcactataat  
aaaatagcttaatacaatattttgttaact tcatgtttaatgcagtgtccctctgtgaagataaattaa  
gcaagaaaaatgaaggcatgtgtktatctttaaaatgaaaatgktttktattcagactaaacttactgccc  
ttctcangggagctaaaattacttactaccactttataatcatctcataaaagatttactctttcca  
gttgc (SEQ ID No. 49)

FIG. 5A

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**Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS**

End intron 3

Ref. No. 21087.0019U3

aaaatgcctg gtttcagaa gggggtycc ataggcttt tggtcagggggngtaar gaggnatt  
gaagctgggg actgttnggg ggaacccnctgttccttg gagcttaant tanggagcng gtcaggggg  
gaaanggagggcttaatn ctgtnanagg nttnaaaaaa aaaaaaaaaa ntccnnggctggtnngggt  
ggggngggg gaaaggcca agaaaaaaaaa aaaaaatggtnnnnnnnn ttaacattt ccaatgtggg  
aaaaaaaaagca aattaataaagagcagtca agaagttgga gaagatttagt ctcaaaacag  
aaaagaagatggtaactggc anctgtacca aaaagaacag aagagtttag gcagctgatggttgagaatg  
gaccccccga gctgtccaat gcacagactt gtctttgaaaaaaaaagcga tagaatgtta aaccacccat  
ctcatcatat atctaggacttttagcacaag gattgttgcc ataagaatga agctttaga  
gtgatttcttaggaatgga cacaccaatt aactgtctcc tggccccacc ttgtatgtttcttcacag  
(SEQ ID No. 50)

### Exon 4

CAATGCACTG ACGGATATGA GTGGGATCCT GTGAGACAGC AATGCAAAG (SEQ ID No. 51)

complete intron 4

gtgagccagcttcaaaggacttccatgtgatcattgccttctgtccatctcttgcactttccctgt  
ctgtgttataccaaaaaggcatgagcattatatttacatgtttgattttccctttagaaraattccctgacttat  
tttattactqaccacaaq (SEQ ID No. 52)

### Exon 5

ATATTGATGA ATGTGACATT GTCCCAGACG CTTGTAAAGG TGGAATGAAG TGTGTCAACC  
 ACTATGGAGG ATACCTCTGC CTTCCGAAAA CAGCCCAGAT TATTGTCAAT AATGAACAGC  
 CTCAGCAGGA AACACAACCA GCAGAAAGGAA CCTCAGGGC AACCACCGGG GTTGTAGCTG  
 CCAGCAGCAT GGCAACCAGT GGAGTGTTCG CCGGGGGTGG TTTTGTGGCC AGTGCCTGCTG  
 CAGTCGCAGG CCCTGAAATG CAGACTGGCC GAAATAACTT TGTCTATCCGG CGGAACCCAG  
 CTGACCCCTCA GCGCATTCCC TCCAACCCCTT CCCACCGTAT CCAGTGTGCA GCAGGCTACG  
 AGCAAAGTGA ACACAACGTG TGCCAAG (SEQ ID No. 53)

Start intron 5

Gtaggaataacttatttcctaaccattgaaagctgcaggagaacccatgaagcttgaattcgatccacgttq (SEQ ID No. 54)

### End intron 5

Tttggcattctgaaattttgcaatgaaagatttatacawacatgtgccctgtttggaaaaacaacacgc  
tacmgggraacggtaataatattaaagggtgatwacacccagttatgttagatttttagaaatttgcataatgg  
aattatctcaaatacaatataattggatggaaaagcaagtgatcatacaatctattaaaattttacatacaaaac  
aataccatatgttctaatggatgcatccctgtctaacaaaagtgacaaaacatctcagggaaggattcattccta  
ccgagacagtggtagctgatgggtcaaggatgaggatgggtgtgaggcttagctgtatctgaaatgttcttaa  
caaaaacaaaatgagccaagaccaacatgacaaaatgttagcattgttaaatctgagcgtactcactggattt  
gcaaaaatttatttctgaaacacttgaataatttataattttaaacatttccatgcaagaacattataaactttt  
aagaataaaagtwwaatttagctttagaagtggcmaatggargaaatcaacatcttcacaactgacaatyyt  
tytstgtgctwgyatgtctswgacag (SEQ ID No. 55)

### Exon 6

ACATAGACGA GTGCACTGCA GGGACGCACTGTAGAGC AGACCAAGTG TGCATCAATT  
TACGGGGATC CTTTGCATGT CAGTGCCCTC CTGGATATCA GAAGCGAGGG GAGCAGTGCCTAG  
(SEQ ID No. 56)

FIG. 5B

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## Title: MACULAR DEGENERATION

## DIAGNOSTICS AND THERAPEUTICS

Inventors: Stone *et al.* Ref. No. 21087.0019U3

Start intron 6

End intron 6

### Exon 7

ACATAGATGA ATGTACCATC CCTCCATATT GCCACCAAAG ATGCGTGAAT ACACCCAGGCT  
CATTTTATTG CCAGTGCAGT CCTGGGTTTC AATTGGCAGC AAACAACTAT ACCTGCGTAG  
(SEQ ID No. 59)

Start intron 7

Gtaaggcctttggagaacttgcgtatctgtcttcacaaaaggaaacccatgccatgtgcagtatccatgtttctatgttcttgcgtaaaatagttccatgcacttcccttcagcaatcataaagytgcaggagaacccatgagcttgcattccggatccacgtgg (SEQ ID No. 60)

End intron 7

aaactttccactgaaagtgcattctgattttacatgccttttgccttcagaaatgcataatgtcacgg  
: cacaacaattaaaagattggcatgacatggaaaaaa:tctagttrggaaaatgcctttcaacaatatt  
t: cagtgccttagaagcattgcaaaactccgtatgggtctcaaaggctatgttataattgtatggaaattt  
acagaacccattnaaaaagttataaa:atagccacagataaatctccagtagccagcattgcctgaagaagac  
catatccagtataagttgttawcaattattttagaaattggcattttgtwtcttgaaccaacaaaagaa  
aaatccgaat:mccggaaktgttatatttttagaagcattaaattcccttgganagattnatcacacatcnna  
ctaactgtcattcctagaaaaatattcggatttccnnaagaagtatatgacagacgctttagttgttccc  
acaaaatgtanaccnnaatggatgttctccagtgagctctgcaggcataattcagctaggaaattactca  
tttgtcagcagatgacgttagtacaaaagagaaggatgtttaagtstaymtatmtgtgttatay  
tatacatatacaymwymtatayatamratttttcwag (SEQ ID No. 61)

## Exon 8

ATATAATGA ATGTGATGCC AGCAATCAAT GTGCTCAGCA GTGCTACAAAC ATTCTGGTT  
CATTCATCTG TCAGTGCAAT CAAGGATATG AGCTAAGCAG TGACAGGCTC AACTGTGAAG  
(SEQ ID No. 62)

FIG.5C

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## Title: MACULAR DEGENERATION DIAGNOSTICS AND THERAPEUTICS

Start intron 8

Inventors: Stone *et al.* Ref. No. 21087.001903

Ref. No. 21087.001903

End intron 8

Aaaaaatataatgtgtgtgtgtgtgtgtgttatattaaacccagncaactaaaaatgt  
gccaagtcacacagtcgcaggaataggacaanaagccagatcttttatataataggtta,gatataattt  
ttcctccttanaatataaataa,tttaatttatataattttatataatagatattttaaatcttataattt  
atataatataattttatataatataatccaaagtagtggcacaactttcaactctgtgtcctt  
tctcttgcataattcaacag (SEQ ID No. 64)

### Exon 9

ACATTGATGA ATGCAGAAC C TCAAGCTACC TGTGTCAATA TCAATGTGTC AATGAACTG GGAAATTCTC  
ATGTATGTGC CCCCAGGGAT ACCAAGTGGT GAGAAGTAGA ACATGTCAAG  
(SEQ ID No. 65)

Start intron 9

gtaaaggtttattttttttcatatgttagtgtttagtttagccaggaaagagacaagaggaagttataggatt  
ctccatagacttcattttccacttcaatatacaatatttaagctntttccctgtcatcataaaaata  
tatacatctcataaagaggggattctatgtaangccacntttcgtccttaaaagataataattttata  
aatatttgatatgtattctatgtaacctacatcatctnttgagatacatctcaatcatccactgaaaaaga  
ttcagttattaaaangttcacctgtgagttgagttanagcataagctcaatatggagttaaacatacctc  
catccagctttagccctctaaaacncanggattataaattgcgtaaaaatgttaggtgctaaaaaagtcc  
aatatgtgtaaaatatagttgaaatattttagagaaaactactagccccaaaatagctaattac  
gtttcaaaataaaccattcagatcttccacccatatacgtaaaggaaagaggcagctccccacaatggatga  
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ttaacaggggaaaggactgtcanataaaacccaaaacgtaaaaatccctggaaaagctagtncaaacncttaat  
ttacncaaagcaccaaaagaataaaaatgaccaancattgacanaaaacccatgttgaatccagctccactgt  
ttcagctgcncatnttgaacaaaattatcaaactactntgagccctcagntccctcatgttggaaaaggagtt  
ggggaaatttagggaaatancatncntaaaaatanttgtaaactataaagcttgcaggtcaaggggaaaa  
atnnaatttac (SEQ ID No. 66)

FIG.5D

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Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS

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End of intron 9

Agccttttccttaacttcttttccttacagtctaaattgtctatgaggtggaaaccttcata  
gtttcactttcctgtgtgtcttcgtggacagtataatccactcccagcatgttcactgaaaccag  
atttctagccttacccatccaaagttctgaaagagatgataagctgcctccatagttatgtttctgaa  
tttctcagcttggaaaggccctccctgcccagccatggaaactccatctaaccaccacccttcaaggccacgtt  
gagatgccttccttcctcagccctccataatccccctggcaaaattacccaactctgtctccatgc  
gtataacttatctatcttacttaattccattttactttctaagtaatcatatacacattccctcaattataat  
gtccctgtatgacaagaactgggttttaactttatataaggcagagtcaactggtaacattggggttgaattca  
acagatgaacaataggcgttataatcatgaaatgacacatataatctgcctagaatgtctcagct  
gtctgtcctgaattcaatacatgaaacacccagtctgtctaaaagcagggttgaacacagttccagatgct  
cacaccccttccttgcacaaacagaatctgccttatgatattagatcaggcatttagtttagta  
aactcttgcaccctacatctctacag (SEQ ID No. 67)

Exon 10

ATATAATGA GTGTGAGACC ACAATGAAT GCGGGAGGA TGAAATGTGT TGGAATTATC  
ATGGCGGCTT CCGTTGTTAT CCACGAAATC CTTGTCAAGA TCCCTACATT CTAACACCAAG AGAA  
(SEQ ID No. 68)

intron 10

gtaagaaaaa tcagaacttt tgaaagttag gatttctgg tcttaccaagccaaactgct gaatactttt  
gtttgtctct gcag  
(SEQ ID No. 69)

Exon 11

CCGATGTGTT TGCCCAAGTCT CAAATGCCAT GTGCCGAGAA CTGCCCAAGT CAATAGTCTA  
CAAATACATG AGCATCCGAT CTGATAGGTC TG GCCATCA GACATCTTCC AGATACAGGC  
CACAACTATT TATGCCAACA CCATCAATAC TTTTGGATT AAATCTGGAA ATGAAAATGG  
AGAGTTCTAC CTACGA (SEQ ID No. 70)

Start intron 11

gtaagtatcc tgaaggcgc cttaactatt gagaagatg ggagttgtgtgttgg tttttttt  
tggatgtgg tatccacatg tggaaaggaaagcaacattt aaaagtgtcttnatgtgttaggcattgtgt  
gccttccagactacattttcatttattcccttggtaacactgcccagatagatattaatattcatctccatttt  
ttacagaggaaaaagttagatgcaaaaaatgatggatcacttcaatatttttttttttttttttttttttttt  
ctctgtatgatgtgtgaccacaaaaatacaaggccctccatccccccgcctgtgcacccacatacc  
cccaagttaggtatcactatcattgtgttattatatacatagacatacatataattcaaaacccaaaaataa  
ttcctggagctctaaagatgttttgcacatcatgttgcatttttttttttttttttttttttttttttttt  
caccggaaactaaaaactataatgtatgtgaaatgtgttgcacccacccatgtgttgcacatgtgttgc  
ttcttgcacacttctggctggcagatggatgttgcatttttttttttttttttttttttttttttttttt  
aattctgcattttcccttcacactgtctctgggttatcttgcatttttttttttttttttttttttttt  
aactggctcccgtaacttccatgtatcccttgcatttttttttttttttttttttttttttttttttt  
ccagtctccctacaaaatctcagcaagtttgcatttttttttttttttttttttttttttttttttt  
ttcatgtgaaaatgtatggcacccttaccaaggcaggatgttgcagatgttgcatttttttttttttt  
tcattccagttac SEQ ID No. 71)

End intron 11

Tcccttttttttcttcaaaaaggnaaccnatggcccaagntgnaaaaaaaaggccncttgnntt  
ccaggtttaaaaatttccnattttccctwaagtttagktttggaaaggccccacttcnccannaaaaggaa  
aaaaaatgnttacmaanaggggggattcaaaacnaaaaacttttttttttttttttttttttttttttt  
acttggagctaatgtactgtttagacaaggataagagccaaagaagagatgttgcacccatgttgc  
ggcttttttataccaccccttcaaaaatttctcccttcaatttttttttttttttttttttttttttt  
aatccaagaaaatttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttctt  
(SEQ ID No. 72)

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Title: MACULAR DEGENERATION  
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Exon 12

CAAACAAGTC CTGTAAGTGC AATGCTTGTG CTCGTGAAGT CATTATCAGG ACCAAGAGAA  
CATATCGTGG ACCTGGAGAT GCTGACAGTC AGCACTATAG GGACCTTCGG CACAAGCTCT  
GTGTTAAGAT TGACAATAAT AGTGGGGCCA TTTTCATTT AG (SEQ ID No. 73)

3' utr

tcttttctaa gagtcaacca caggcattta agtcagccaa agaatattgt taccttaaag  
caactatTTta ttatagata tatctagtgc atctacatct ctatactgtta cactcaccca  
taacaaacaa ttacaccatg gtataaagtg ggcatttaat atgtaaagat tcaaagttt  
tcttttattac tatatgtaaattagacatta atccactaaa ctggcttct tcaagagagc  
taagtataca ctatctggtg aaacttggat tctttcctat aaaagtggga ccaagcaatg  
atgatcttct gtggtgctta agggaaactta cttagagctcc actaacagtc tcataaggag  
gcagccatca taaccattga atagcatgca agggtaagaatgagttttta actgctttgt  
aagaaaaatgg aaaaggctaa taaagatatatTTCTTtaga aaatggggat ctgccatatt  
tgtgttgggt tttatTTTcatatccagcct aaagggtgggt gtttattata tagtaataaaa  
tcattgtgtacaacatgct ggttctgtta gggattttt aattttgtca gaaatttttagattgtgaata  
ttttgtaaaa aacagtaagc aaaattttcc agaattcccaaaatgaacca gataccccct agaaaattat  
actattgaga aatctatggggaggatatga gaaaataaaat tccttctaaa ccacattgg  
actgacctgaagaagcaaac tcggaaaata taataacatc cctgaattca ggcattcacaagatgcagaa  
caaaatggat aaaaggattt tcactggaga agttttaaatttctaagtaaa atttaaatcc taacacttc  
ctaatttata actaaaatttctcatcttcg tacttgatgc tcacagagga agaaaatgt  
gatggtttttattcctggca tccagagtga cagtgaactt aagcaaattt ccctcctacccaattctatg  
gaatattttta tacgtctcct tggtaaaat ctgactgctttactttgatg tatcatattt ttaaataaaaa  
(SEQ ID No. 74)

FIG.5F

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Title: MACULAR DEGENERATION  
DIAGNOSTICS AND THERAPEUTICS

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M LKALFLTMLTL ALVKSQDTE ETITYTQCTD GYEWPVRQQ  
CKDIDECDIV PDACKGGMKC VNHYGGYLCL PKTAQIIVNN EQPQQETQPA  
EGTSGATTGV VAASSMATSG VLPGGGFVAS AAAVAGPEMQ TGRNNFVIRR  
NPADPQRIPS NPSHRIQCAA GYEQSEHNVC QDIDECTAGT HNCRADQVCI  
NLRGSFACQC PPGYQKRGEQ CVDIDECTIP PYCHQRCVNT PGSFYCQCSP  
GFQLAANNYT CVDINECDAS NQCAQQCYNI LGSFICQCNQ GYELSSDRLN  
CEDIDECRTS SYLCQYQCVN EPGKFSCMCP QGYQVVRSSRT CQDINECETT  
NECREDEMCW NYHGGFRCYP RNPCQDPYIL TPENRCVCPV SNAMCRELPQ  
SIVYKYMSIR SDRSVPSDIF QIQATTIYAN TINTFRIKSG NENGEFYLRQ  
TSPVSAMLVL VKSLSGPREH IVDLEMLTVS SIGTFRTSSV LRLTIIVGPF  
SF (SEQ ID NO.: 2)

FIG.6